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COLPOCEPHALY IN ADULTS: A RARE CASE REPORT

Neurology

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ABSTRACT

BACKGROUND - Colpocephaly is a congenital abnormality in the ventricular system of the brain. The radiological diagnosis is usually made in the perinatal period and later presages intellectual disability. Adult cases of newly diagnosed colpocephaly have only rarely been reported.

CASE SUMMARY – The patient is a 15-year-old right-handed female presented to the emergency room for evaluation after a series of falls along with a noticeable foot deformity on left side Questioning revealed lifelong gait instability. She was alert, appropriately oriented and had normal language function. Mental status review showed a flattened affect and deficits in recent memory Non-contrast brain MRI revealed massive symmetrical dilation of the posterior lateral and the third ventricles, thin parieto-occipital white matter and partial agenesis of the corpus callosum. She was diagnosed with congenital colpocephaly because of the radiological findings. She came to a rehabilitation facility for gait training and management of the foot deformity.

CONCLUSION - When encountering ventriculomegaly in an adult, one must distinguish between the different forms of obstructive and nonobstructive ventriculomegaly. We propose that colpocephaly be considered in the differential for adults with non-obstructive ventriculomegaly. When secondary causes have been ruled out, distinguishing between colpocephaly and NPH can be done through a careful history, physical examination and evaluation of the radiological characteristics. Doing so can prevent the iatrogenic risk associated with surgical shunting.

KEYWORDS

colpocephaly, NPH, Ventriculomegaly.

INTRODUCTION

Colpocephaly is a widely recognised paediatric diagnosis, with only two other adult cases described in medical literature[1,2] It is a congenital form of ventriculomegaly that was first described by Benda in 1941[3]. The original patient was a 3-year-old boy with intellectual disability, paralysis and seizures—many of the clinical features now commonly associated with the disorder. He died at the age of 10. Autopsy revealed enlarged lateral ventricles, thin and undifferentiated occipital lobes, absent corpus callosum, macrogyria and microgyria.

CASE REPORT

A 15-year-old female presented to the emergency room for evaluation after a series of falls. Questioning revealed lifelong gait instability. She suffered a major mechanical fall 1 month prior to presentation, resulting in minor head trauma. Since then, she reported daily headaches, worsening gait instability and increasing frequency of falls. Her medical history was significant for poor vision, and lifelong clumsiness which were said to be sequelae from congenital toxoplasmosis. Growing up, she had a reading learning disability; however, she graduated from high school with average grades. At the time of presentation, she was admitted to difficulty doing housework. She was alert, appropriately oriented and had normal language function. Mental status review showed a flattened affect and deficits in recent memory. She had bilateral vitreous opacities and cataracts. Her right pupil was unreactive, while the left pupil was only sluggishly reactive. Vision was minimally present on the left. She had normal strength, sensation and appendicular coordination. Alternating movements were slow and reflexes decreased symmetrically. Gait was wide-based and unsteady.

INVESTIGATIONS

Serum sodium content was 116 mmol/l. Work up was consistent with the syndrome of inappropriate antidiuretic hormone secretion (SIADH). Noncontrast CT of the head showed massive dilation of the lateral and third ventricles. The fourth ventricle was relatively normal with no evidence of obstruction. Multiple punctate calcific densities were scattered within the brain parenchyma, consistent with congenital toxoplasmosis (figure 1). Non-contrast brain MRI revealed massive symmetrical dilation of the posterior lateral and the third ventricles, thin parieto-occipital white matter and partial agenesis of the corpus callosum. Multiple thin crescentic subdural haematomas—deemed to be subacute— were noted on fluid-attenuated inversion recovery MRI (figure 2). She was diagnosed with congenital colpocephaly because of the radiological findings of (1) nonobstructive ventriculomegaly (2) disproportionate dilation of the occipital horns compared to the frontal horns and (3) partial agenesis of the corpus callosum. Brain calcifications and ventriculomegaly, combined with a history of congenital blindness, were all consistent with the history of congenital toxoplasmosis. The recently decompensated gait was attributed to SIADH, which likely developed as a result of traumatic brain injury suffered in a recent fall.

OUTCOME AND FOLLOW-UP

Hyponatraemia improved with conservative management and she was discharged to a rehabilitation facility for continued gait training. Her gait had improved, but she required assistance with the activities of daily living because of the coexistent foot deformity.

DISCUSSION

Colpocephaly is a widely recognised paediatric diagnosis, with only two other adult cases described in medical literature[1,2] It is a congenital form of ventriculomegaly that was first described by Benda in 1941[3] The original patient was a 3-year-old boy with intellectual disability, paralysis and seizures—many of the clinical features now commonly associated with the disorder. He died at the age of 10. Autopsy revealed enlarged lateral ventricles, thin and undifferentiated occipital lobes, absent corpus callosum, macrogyria and microgyria. Colpocephaly can result from a wide range of congenital insults.

Chromosomal abnormalities, maternal toxin exposure, anoxic encephalopathy and intrauterine infection, such as toxoplasmosis, have been described as potential causes. Just before the fifth month of life, the fetus undergoes a stage of relative hydrocephalus that is normally abolished by the migration of glial cells and growth of the surrounding white matter and corpus callosum [4,5]Any intrauterine insult that interferes with this maturation process can theoretically cause the retained fetal ventricular configuration that defines colpocephaly. Radiologically, colpocephaly is characterised by disproportionate dilation of the occipital horns and is commonly associated with full or partial agenesis of the corpus callosum[6]. In fact, both previously published adult cases were associated with complete agenesis of the corpus callosum, while our patient had partial agenesis of the corpus callosum.

Other radiological associations include microcephaly, myelocele, macrogyria, microgyria, schizencephaly, lissencephaly, pachygyria, cerebellar atrophy and optic nerve atrophy[7]. In contrast, idiopathic NPH is an adult onset form of progressive ventriculomegaly. It was originally described as causing the clinical triad of disabling dementia, gait disturbance and urinary incontinence[8]. The diagnosis of

"probable" NPH is made by demonstrating at least two of the three features in the clinical triad, a normal cerebrospinal fluid pressure at lumbar puncture and the presence of non-obstructive dilation of the ventricular system that is disproportionate to the level of brain atrophy[9,10] . Early in the course, there may be transependymal flow in the periventricular white matter or dilation of the temporal horns. With disease progression, the lateral ventricles are affected, culminating in ballooning of the frontal horns. Quantitative measures include the Evans index, callosal angle, aqueductal flow rate, apparent diffusion coefficient and intracranial compliance index [11,12,13,14] Colpocephaly can also be assessed quantitatively.

The posterior to anterior ratio (P/A ratio) was originally described by Noorani, Bodensteiner and Barnes as a measure of occipital horn enlargement. To calculate, the maximal width of the occipital horn is divided by the maximal width of the anterior horn of the lateral ventricle (figure 3). Disproportionate enlargement of the occipital horns was defined as a P/A ratio ≥3, and was found positive in 7 of the 14 patients in the original cohort. In the remaining cases, diagnosis of colpocephaly was based on the recognition of the characteristic configuration of the lateral ventricles. In the right clinical setting, a P/A ratio ≥3 is highly specific for colpocephaly and may be used as a diagnostic tool to distinguish colpocephaly from NPH. Because of the low sensitivity, one must also take into account the history and physical examination findings. When encountering ventriculomegaly in an adult, one must distinguish between the different forms of obstructive and nonobstructive ventriculomegaly. We propose that colpocephaly be considered in the differential for adults with non-obstructive ventriculomegaly. When secondary causes have been ruled out, distinguishing between colpocephaly and NPH can be done through a careful history, physical examination and evaluation of the radiological characteristics. Doing so can prevent the iatrogenic risk associated with surgical shunting [15].

CONCLUSION

Colpocephaly is a form of congenital ventriculomegaly, characterised by non-obstructive ventriculomegaly, disproportionate dilation of the occipital horns and often associated with partial or full agenesis of the corpus callosum. Clinically, colpocephaly is associated with motor abnormalities, intellectual disability, vision problems and seizures. When encountered radiologically, colpocephaly must be distinguished from normal pressure hydrocephalus (NPH). The posterior to anterior ratio ratio may aid in distinguishing colpocephaly from NPH.

Conflict of interest - The authors declare no conflict of interest whatsoever arising out of the publication of this manuscript.

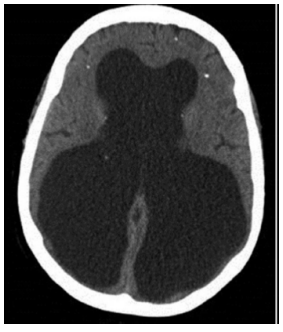

Figure 1. CT scan demonstrating ventriculomegaly and punctuate calcifications within the Brain parenchyma , consistent with given history of congenital Toxoplasmosis.

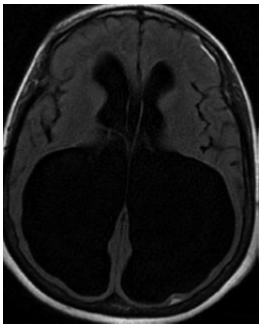

Figure 2. MRI Brain showing massive dilatation of the occipital horns and of third ventricle.

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